Pharma Ontology: Creating a Patient-Centric Ontology for Translational Medicine

Christine Denney¹, Colin Batchelor², Olivier Bodenreider³, Sam Cheng⁴, John Hart⁴, John Hill⁴, John Madden⁵, Mark Musen⁶, Elgar Pichler⁷, Matthias Samwald⁸, Sándor Szalma⁹, Lynn Schriml¹⁰, David Sedlock¹¹, Larisa Soldatova¹², Koji Sonoda¹³, David Statham¹¹, Trish Whetzel⁶, Elizabeth Wu¹⁴, Susie Stephens¹

¹Eli Lilly, Indianapolis, IN, USA; ²Royal Society of Chemistry, Cambridge, UK; ³National Library of Medicine, Bethesda, MD, USA; ⁴Boehringer Ingelheim, Ridgefield, CT, USA; ⁵Duke University, Durham, NC, USA; ⁶Stanford University, Stanford, CA, USA; ⁷AstraZeneca, Waltham, MA, USA; ⁸DERI Galway, Galway, Ireland & KLI, Austria ⁹Centocor R&D, San Diego, CA, USA; ¹⁰University of Maryland, College Park, MD, USA; ¹¹Millennium Pharmaceuticals, Cambridge, MA, USA; ¹²University of Aberystwyth, Aberystwyth, UK; ¹³Amgen, Thousand Oaks, CA, USA; ¹⁴Alzheimer's Research Forum, Cambridge, MA, USA

Abstract

We, participants in the Pharma Ontology activity of the World Wide Web Consortium's Semantic Web for Health Care and Life Sciences Interest Group (http://esw.w3.org/topic/HCLSIG) and members of the National Center for Biomedical Ontology (http://bioontology.org/), are developing a highlevel, patient-centric ontology for translational medicine which will draw on existing domain ontologies and allow the integration of data throughout the drug development process.

Introduction

The pharmaceutical industry has historically focused on the development of novel blockbuster drugs. There is now an increasing focus on personalized medicines, requiring the right patients to receive the right drug at the right dose. In order to develop a tailored drug, manufacturers need to identify biomarkers that will indicate how a given patient will respond to a particular treatment. Biomarkers can also be used to demonstrate the comparative effectiveness of drugs, which is increasingly required by payers. Such translational medicine strategies require that traditionally separate data sets from early drug discovery through to patients in the clinical setting be integrated, and presented, queried and analyzed collectively. Ontologies can be used to drive such data integration and analysis, however, at present few ontologies exist that bridge genomics, chemistry and the medical domain.

The Pharma Ontology, an application ontology that bridges the diverse areas of translational medicine, draws on existing domain ontologies where appropriate and will provide a framework centered on less than 50 types of entities.

Goals

The Pharma Ontology will facilitate data integration from diverse areas of translational medicine such as research, hypothesis management, formulation, clinical trials, and clinical research. It will serve as a template for further ontology enabling scientists development, to interesting and currently difficult questions more easily, especially those about data that are typically hosted by different functional areas. The ontology will provide a framework for the modeling of patientcentric information, which is essential for tailoring drugs.

Methodology

We have identified a set of 17 roles played by people across health care and the life sciences and collected (1) relevant questions, (2) the entities that those questions involve, and (3) applicable extant domain ontologies. Types of entities include: disease, drug, patient, target, gene, risk, pathway, population, compound, phenotype, and treatment.

Next steps will involve identifying use cases based on those questions, determining which entities to build into the ontology and aligning them with BFO,² an upper-level ontology, to aid interoperability between domain ontologies. We will use one use case to test the Pharma Ontology by building a data integration application based on it.

Conclusion

This project seeks to develop a patient-centric application ontology for translational medicine, as a collaborative effort between groups in industry and academia. The presentation will highlight our methodology, work to date, and future steps.

- 1. http://esw.w3.org/topic/HCLSIG/PharmaOntology/Roles
- 2. http://www.ifomis.org/bfo